

Join us for URBC Virtual Research Week: July 6th-9th 2020



We are hosting a **virtual research** week this year! The event will occur July 6th-9th, leading up to the annual BDSRA meeting. You are invited to participate virtually in our research projects during this week. In addition to research visits, we will be hosting virtual coffee hours during meeting breaks on July 10-12. Watch our Facebook page for updated information as well as additional activities we will participate in throughout the BDSRA meeting. We look forward to seeing many of you virtually this July!

Who can Participate?

All families of individuals with a NCL diagnosis are invited to participate.

How to Schedule a Visit

Study visits can be scheduled for Mon-Thursday during the week of the BDSRA conference (July 6th-9th). Call (585) 275-4762 or email Batten@urmc.rochester.edu to learn more or to schedule a virtual research visit.

Research in the Midst of COVID-19

The URBC is making every effort to allow families to continue their participation in University of Rochester research projects. Your and your family's safety will always be our priority. Therefore, we have made a temporary shift to virtual visits that can be conducted from the safety and comfort of your own home.

Telehealth in Batten Disease – Past, Present, & Future

Past: The story begins... telemedicine research project

Nearly 10 years ago, researchers at the URBC set out to learn if some of our core study activities could be conducted via 'telemedicine', when the researcher and the research participant are in different physical locations. The project was inspired by a desire to increase access to research opportunities for families who were geographically spread throughout the country, and to reduce the burdens associated with travel to distant research sites. Travel-related burdens included the time, distance and costs involved with travel, lost time from work, and the challenges of traveling with a child who has disabilities. Our telemedicine project resulted in two publications. The first, "[Quantitative telemedicine ratings in Batten disease](#)" demonstrated that it is possible to administer the Unified Batten Disease Rating Scale (UBDRS) through telemedicine. The UBDRS is a Batten Disease-specific neurologic exam and rating scale to assess Batten symptoms and disease severity. The second paper, "[Remote Assessment of Cognitive Function in Juvenile Neuronal Ceroid Lipofuscinosis](#)" demonstrated that it is also possible to conduct telemedicine-based cognitive evaluations of children with Batten disease. In both studies, we found that telemedicine research visits were possible, practical, and resulted in high-quality data.

Present: Research and virtual visits in the midst of COVID-19

When we began our telemedicine project, we could not have anticipated another reason for remote study visits – the current COVID-19 pandemic. However, our past experience has made it possible for families to continue participating in URBC research studies and clinical care during this time. Last month we began virtual research visits, which can be conducted from the safety and comfort of your own home. Tele-research virtual visits use software that protects your privacy and confidentiality. At the University of Rochester, we use an app called "Zoom" for tele-research visits. Zoom works similarly to video calls on FaceTime or Skype. We use Zoom because it is compliant with health privacy laws (called HIPAA), and protects your personal, private health information during the research visit.

Future: Tele-research and tele-health – where do we go next?

The URBC is dedicated to advancing research and clinical care for all forms of Batten disease. We look forward to the day when we can welcome you back to Rochester and see you again in person at BDSRA meetings and other events during the year. There will also be important research and clinical activities that are best conducted in-person, or are only possible at a research or clinical center. We deeply value the time we spend with affected individuals and their families, and we miss seeing all of you! However, we see opportunities to continue expanding tele-research to reach individuals who are unable to travel, to reduce the cost and time related to participation in research, and to diversify the types of research available to you (such as remote and more frequent monitoring of symptoms in the everyday environment).

Translational Research Conference

URBC members presented their research at the Translational Research Conference for the Management of NCLs hosted by Our Promise To Nicholas Foundation (Chicago, IL, November 7-8, 2019). Margaux Masten and Dr. Mink presented “Natural History Studies in the NCLs: One Size Does Not Fit All” and Dr. Augustine presented “Clinical Trial Readiness: Moving from Natural History to Outcome Measures and Endpoints”. It was a wonderful opportunity to interact with many Batten disease stakeholders including leaders of family foundations, NCL researchers, and representatives of companies that are developing treatments for the NCLs.



Cross-validation of the Vineland-3 with Independent Assessments of Cognition and Adaptive Skills in CLN3 Disease:

In this project, we found that there was good agreement among three independent methods to assess affected children’s cognition and everyday skills: parent ratings, clinician exam (UBDRS), and neurocognitive testing.

CLN1 Disease Natural History Data: Prospective and Retrospective Analysis:

We studied CLN1 disease natural history using two strategies: prospective, in-person evaluations and retrospective, medical chart review. Information from both studies showed no pattern in the age-at-onset of core features of the NCLs for any of the age-based groups (infantile, late infantile, or juvenile). Based on information from the prospective study, CLN1 disease appears to progress rapidly in all age-based groups. Overall, we found that CLN1 disease is more variable than other NCLs. Considering its rarity, a combined prospective and retrospective approach to studying CLN1 disease will be beneficial.



New Publication: CLN3 Staging System

We recently developed and tested a clinical staging system for CLN3 disease (CLN3 Staging System; CLN3SS). Because CLN3 disease unfolds more slowly than other forms of Batten disease, division into groups based on discrete and meaningful clinical features is likely to be useful. In the CLN3SS, an individual is in stage 0 if they have genetically confirmed CLN3 disease but have not yet developed symptoms. Once there is evidence of vision loss, an individual enters stage 1. Stage 2 represents individuals who have vision loss and have experienced their first CLN3 disease-related seizure. Stage 3 represents individuals who have lost the ability to walk independently. Our goal is for this staging system to be useful for dividing individuals into groups that will lead to more efficient clinical trials in CLN3 disease.

<https://pubmed.ncbi.nlm.nih.gov/32300063/>

WORLD Symposium

URBC Members presented seven posters on Batten disease research at the 16th Annual WORLD Symposium in Orlando, Florida (Feb. 10-13).

American Academy of Neurology Virtual Conference

The URBC presented data from three projects at the virtual American Academy of Neurology conference.

Age-at-onset and Time-to-event of Core Features in CLN3 (Batten) Disease, Masten et al.: <https://slide-us.ctimeetingtech.com/aan2020/attendee/eposter/poster/3931?q=masten>

Cross-sectional and Longitudinal Natural History of CLN3 Disease Progression, Masten et al.: <https://slide-us.ctimeetingtech.com/aan2020/attendee/eposter/poster/2156?q=masten>

Quantitative Gait Analysis in CLN3 Disease, Zimmerman et al.: <https://slide-us.ctimeetingtech.com/aan2020/attendee/eposter/browse/gallery?q=zimmerman>

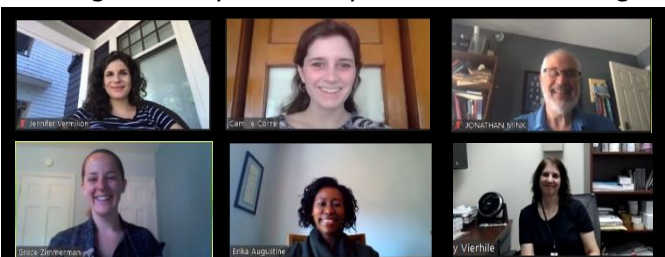
Sleep Study

BDSRA awards grant to UR Batten Center to study sleep in Batten disease

Many parents of children with Batten disease report that their affected children have difficulty falling and staying asleep, that they sleep a lot during the daytime, or that their sleep quality is poor. However, there is little known about the specific sleep patterns of children with Batten disease, or what might contribute to sleeping problems. Through a generous grant from the Batten Disease Support & Research Association, ForeBatten, and Noah’s Hope/Hope for Bridget, the URBC will conduct a new study, titled, “Characterizing Sleep Dysfunction in individuals with NCL Disorders” (Principal Investigator: Heather Adams, PhD). The study will focus on the sleep patterns and the sleep/wake cycle of children with CLN2 and CLN3 disease. It will also examine if the child’s sleep is related to quality of life in affected children and their parental caregivers. Over a two-week period, we will ask parents to complete logs of children’s daily activities and sleep routines, and ask affected children to wear a wristwatch-type activity monitor (like a ‘fitbit’) to measure their movements during daytime/awake and nighttime/sleeping hours. We will also collect saliva samples to measure melatonin levels. Melatonin is a naturally produced hormone that helps to cue the body to prepare for sleep.

Due to COVID-19 related safety precautions for research activities, the URBC is delaying the start of this study. We have notified the BDSRA of our request to delay the study start, and will communicate with BDSRA and the Batten community as a whole when we are able to begin this research study.

The URBC team has been meeting virtually since March to continue our research efforts remotely. We look forward to seeing some of you virtually at the BDSRA meeting!



Study Updates

Clinical Trial Readiness for CLN3 Disease

We are actively enrolling individuals with CLN3 disease for a study focused on finding optimal ways to measure the signs and symptoms of CLN3 disease. The purpose to improve the future clinical trial designs. During visits for this study, participants will be asked to complete an MRI, an eye assessment, a neurological evaluation (UBDRS), a cognitive evaluation and an assessment of walking. Visits typically occur in Rochester, NY over two days. We have adapted our protocol so that we are able to conduct portions of the study remotely. It is possible to enroll in the study now and complete your first research visit from your home.

Natural History Study

We invite families with children who have any form of NCL disorders to participate in our ongoing natural history study. The focus of this study is to better understand how NCL disorders progress. Many families participate in this study year after year, which helps us learn how symptoms change over time. Understanding the progression of NCL disorders is important for measuring the effects of future therapies. Typically, study visits occur in Rochester, NY at the URBC or at the annual BDSRA meetings. Currently, all study activities are being conducted remotely. Please be sure to contact us if you would like to schedule a visit during the week of the BDSRA Annual meeting or any time this summer!

Contact Registry

Sign up to be contacted about future research and receive updates about the University of Rochester Batten Center. You can sign up on our website: <https://www.urmc.rochester.edu/neurology/batten-disease-center.aspx>. Individuals with any form of Batten disease are eligible to enroll.

Medical Records Study

The goal of this study is to understand more about the symptoms of NCL disorders and how those symptoms change over time. For this study, our research team will request healthcare and school records from your child's providers. Individuals with any form of Batten disease, who are living or deceased, are eligible to enroll.

Meet the Team



Grace Zimmerman is a graduate student at the University of Rochester, and a clinical coordinator for the URBC. She is interested in understanding how walking changes as Batten disease progresses. Working with Dr. Augustine, Grace has led the

development of our protocol for obtaining data on walking and analyzed data that was presented at several conferences this year.



Margaux Masten is a recent honors graduate of the University of Rochester with a Bachelor's Degree in Epidemiology. She is now working with the URBC as a study coordinator and continuing her work with Dr. Mink and Dr. Augustine to characterize and

examine the public health impacts of the NCLs.

BDSRA Annual Meeting



We are excited to offer the opportunity to participate in virtual research the week of the Annual BDSRA meeting. Visits will be done through a secure video conferencing Zoom software. Please contact us by phone at (585) 275-4762 or by email if you would like to participate!

In addition to research visits, we will be hosting virtual coffee hours during meeting breaks July 10th to the 12th. Keep an eye on our Facebook page for updated information on timing and topics as well as additional activities we will participate in throughout the meeting. We look forward to seeing many of you virtually at the meeting!

We thank all of the children and families who participate in our research – your contributions help to advance knowledge about Batten disease!

We love to hear from you! Please contact us with your questions and comments. With your permission, we may post answers to your questions on our Facebook page and in upcoming issues of our newsletter.



Phone: (585) 275-4762



E-mail: batten@urmc.rochester.edu



Web: www.rochesterbatten.urmc.edu



<https://www.facebook.com/URMCBattenCenter>